National Registry of Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions (GenTAC)

March 2013

Message from Dr. Kim Eagle, Steering Committee Chair

This is the first of periodic newsletters about the GenTAC Registry and the exciting progress being made. We have over 3,330 people enrolled, and expect to have 3,500 people enrolled by September 2013, when enrollment ends.

The National Registry of Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions (GenTAC) was established in 2006 to enable research to advance the clinical management of patients with genetically induced thoracic aortic aneurysms and related cardiovascular complications. It is funded by the National Heart, Lung and Blood Institute (NHLBI) and the National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS). GenTAC developed standard methods for longitudinal clinical data collection and biospecimen collection. Data include confirmed phenotype, genotype, interpretation of images by a central imaging lab, characteristics of patients undergoing surgical repair of aneurysms and their clinical outcomes, and extensive organ system information related to these conditions. Biospecimens include DNA, plasma, immortalized lymphocytes and surgical tissue. These resources are available through an application process to investigators interested in advancing the fundamental understanding of genetic aortic aneurysms and management of afflicted patients.

Taken at the Second GenTAC Thoracic Aortic Summit in 2012

Bill Ravekes and Kathy Holmes

Reed Pyeritz and Meg Cunningham

Jo Grima and Eser Tolunay

Recent Publications

- Kroner BL, Tolunay HE, Basson CT, Pyeritz RE, Holmes KW, Maslen CL, Milewicz DM, Lemaire SA, Hendershot T, Desvigne-Nickens P, Devereux RB, Dietz HC, Song HK, Ringer D, Mitchell M, Weinsaft JW, Ravekes W, Menashe V, Eagle KA; *The National Registry of Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions (GenTAC): Results from phase I and scientific opportunities in phase II.* Am Heart J. 2011;162(4):627-632. PMCID: PMC3190125
- Song HK, Kindem M, Bavaria JE, Dietz HC, Milewicz DM, Devereux RB, Eagle KA, Maslen CL, Kroner BL, Pyeritz RE, Holmes KW, Weinsaft JW, Menashe V, Ravekes W, Lemaire SA; *Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions Consortium: Long-term implications of emergency versus elective proximal aortic surgery in patients with Marfan syndrome in the Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions Consortium Registry.* J Thorac Cardiovasc Surg. 2012;143(2):282-6. PMCID: PMC3260411
- Holmes KW, Maslen CL, Kindem M, Kroner BL, Song HK, Ravekes W, Dietz HC, Weinsaft JW, Roman MJ, Devereux RB, Pyeritz RE, Bavaria J, Milewski K, Milewicz DM, LeMaire SA, Hendershot T, Eagle KA, Tolunay HE, Desvigne-Nickens P, and Silberbach M for the GenTAC Registry Consortium; *GenTAC Registry Report: Gender Differences Among Individuals with Genetically-Triggered Thoracic Aortic Aneurysm and Dissection.* Am J Med Genet (in press).

Abstracts

- The relationship between aortic and PA diameter among individuals with genetic conditions of the thoracic aorta: A report from the NIH GenTAC Registry. Arteriosclerosis, Thrombosis, and Vascular Biology, April 2012.
- The National Registry of Genetically Triggered Thoracic Aortic Aneurysms (GenTAC): Registry Progress and Research Successes. The American Society of Human Genetics, November 2012.
- Utilization and Outcomes of Valve Sparing Aortic Root Replacement in Patients with Marfan Syndrome Enrolled in the National Registry of Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions (GenTAC). The Society of Thoracic Surgeons, January 2013.

Steering Committee Members

Principal Investigators	
Scott A. LeMaire, MD	Baylor College of Medicine
William Ravekes, MD	Johns Hopkins University School of Medicine
Nazli B. McDonnell, MD, PhD	NIA at Harbor Hospital
Cheryl L. Maslen, PhD	Oregon Health & Science University
Ralph V. Shohet, MD	Queen's Medical Center
Reed E. Pyeritz, MD, PhD	University of Pennsylvania School of Medicine
Dianna M. Milewicz, MD, PhD	University of Texas Medical School at Houston
Richard B. Devereux, MD	Weill Cornell Medical College of Cornell University
Core Labs	
Jennifer P. Habashi, MD	Johns Hopkins University Hospital
Federico M. Asch, MD	MedStar Research Institute
Data Coordinating Center	
Barbara L. Kroner, PhD	RTI International
NHLBI	
Eser H. Tolunay, PhD	National Heart, Lung, and Blood Institute
SC Chair	
Kim A. Eagle, MD	University of Michigan

Taken at the Second GenTAC Thoracic Aortic Summit in 2012

Cheryl Maslen and Michael Silberbach

2012 Highlights

- On August 1-2, GenTAC hosted the second Thoracic Aortic Disease Summit. Attended by over 140 people from across the country, it was a great opportunity to share current findings regarding disease pathogenesis, progression, and treatment and to initiate new collaborations. The next GenTAC Summit will be held in Baltimore, MD in 2014 in connection with the 30th Annual National Marfan Foundation Conference.
- Dr. Simon Body received an NIH grant to study the genetic mechanisms of bicuspid aortic valve (BAV), one of GenTAC's eligibility diagnoses.We are very excited to be collaborating with Dr. Body on this groundbreaking research project.

Do you have a research interest in genetically triggered thoracic aortic conditions?

GenTAC makes its collection of medical data and biologic samples available at no cost to qualified investigators. Your work can help determine best practices that advance the clinical management of genetic aortic aneurysms and other cardiovascular conditions.

» To submit a proposal to use GenTAC data or for more information, visit our website: **http://gentac.rti.org.**

A Snapshot of Who is Enrolled in GenTAC

Number of people enrolled: 3332

Biospecimens

Blood:	1924
Saliva:	1159
Tissue:	137

Gender

Male:	1968
Female:	1336

Eligible Diagnosis

Marfan:	852
Turner:	246
Ehlers-Danlos (vascular):	128
Ehlers-Danlos (other):	20
Loeys-Dietz:	92
FBN1, TGFBR mutation:	25
BAV with aortic enlargement or family history:	849
BAV with coarctation:	75
Shprintzen-Goldberg:	22
Familial TAA:	290
Other aneurysm, dissections:	585
Other congenital heart disease:	92

Age

<5	95
5–17	531
18–39	1003
40–69	1525
>69	114

Race

White, non-Hispanic	2695
Black, non-Hispanic	166
Hispanic	259
Asian	126
American Indian/Alaskan native	13
Native Hawaiian/Pac. Islander	42